

【2020年】

<遺伝、染色体異常、先天奇形>

Baynam G. S., Groft S., van der Westhuizen F. H., Gassman S. D., du Plessis K., Coles E. P., Selebatso E., Selebatso M., Gaobinelwe B., Selebatso T., Joel D., Llera V. A., Vorster B. C., Wuebbels B., Djoudalbaye B., Austin C. P., Kumuthini J., Forman J., Kaufmann P., Chipeta J., Gavhed D., Larsson A., Stojiljkovic M., Nordgren A., Roldan E. J. A., Taruscio D., Wong-Rieger D., Nowak K., Bilkey G. A., Easteal S., Bowdin S., Reichardt J. K. V., Beltran S., Kosaki K., van Karnebeek C. D. M., Gong M., Shuyang Z., Mehrian-Shai R., Adams D. R., Puri R. D., Zhang F., Pachter N., Muenke M., Nellaker C., Gahl W. A., Cederroth H., Broley S., Schoonen M., Boycott K. M., Posada M. A call for global action for rare diseases in Africa. *Nat Genet* 2020; 52(1): 21–26. <doi:10.1038/s41588-019-0552-2>. <https://www.ncbi.nlm.nih.gov/pubmed/31873296>

Fujinami-Yokokawa Y., Fujinami K., Kuniyoshi K., Hayashi T., Ueno S., Mizota A., Shinoda K., Arno G., Pontikos N., Yang L., Liu X., Sakuramoto H., Katagiri S., Mizobuchi K., Kominami T., Terasaki H., Nakamura N., Kameya S., Yoshitake K., Miyake Y., Kurihara T., Tsubota K., Miyata H., Iwata T., Tsunoda K., Japan Eye Genetics Consortium. Clinical and Genetic Characteristics of 18 Patients from 13 Japanese Families with CRX-associated retinal disorder: Identification of Genotype-phenotype Association. *Sci Rep* 2020; 10(1): 9531. <doi:10.1038/s41598-020-65737-z>. <https://www.ncbi.nlm.nih.gov/pubmed/32533067>

Fujita H., Sasaki T., Miyamoto T., Akutsu S. N., Sato S., Mori T., Nakabayashi K., Hata K., Suzuki H., Kosaki K., Matsuura S., Matsubara Y., Amagai M., Kubo A. Premature aging syndrome showing random chromosome number instabilities with CDC20 mutation. *Aging Cell* 2020; 19(11): e13251. <doi:10.1111/accel.13251>. <https://www.ncbi.nlm.nih.gov/pubmed/33094908>

Hara-Isono K., Matsubara K., Fuke T., Yamazawa K., Satou K., Murakami N., Saitoh S., Nakabayashi K., Hata K., Ogata T., Fukami M., Kagami M. Genome-wide methylation analysis in Silver-Russell syndrome, Temple syndrome, and Prader-Willi syndrome. *Clin Epigenetics* 2020; 12(1): 159. <doi:10.1186/s13148-020-00949-8>. <https://www.ncbi.nlm.nih.gov/pubmed/33092629>

Hara-Isono K., Matsubara K., Mikami M., Arima T., Ogata T., Fukami M., Kagami M. Assisted reproductive technology represents a possible risk factor for development of

epimutation-mediated imprinting disorders for mothers aged \geq 30 years. *Clin Epigenetics* 2020; 12(1): 111. <doi:10.1186/s13148-020-00900-x>.

<https://www.ncbi.nlm.nih.gov/pubmed/32698867>

Hiraide T., Kataoka M., Suzuki H., Aimi Y., Chiba T., Isobe S., Katsumata Y., Goto S., Kanekura K., Yamada Y., Moriyama H., Kitakata H., Endo J., Yuasa S., Arai Y., Hirose N., Satoh T., Hakamata Y., Sano M., Gamou S., Kosaki K., Fukuda K. Poor outcomes in carriers of the RNF213 variant (p.Arg4810Lys) with pulmonary arterial hypertension. *J Heart Lung Transplant* 2020; 39(2): 103–112. <doi:10.1016/j.healun.2019.08.022>.

<https://www.ncbi.nlm.nih.gov/pubmed/31542298>

Ishizaki-Asami R., Uchida K., Tsuchihashi T., Shibata A., Kodo K., Emoto K., Mikoshiba K., Takahashi T., Yamagishi H. Inositol 1,4,5-trisphosphate receptor 2 as a novel marker of vasculature to delineate processes of cardiopulmonary development. *Dev Biol* 2020; 458(2): 237–245. <doi:10.1016/j.ydbio.2019.11.011>.

<https://www.ncbi.nlm.nih.gov/pubmed/31758944>

Kawamura Y., Toyoda Y., Ohnishi T., Hisatomi R., Higashino T., Nakayama A., Shimizu S., Yanagi M., Kamimaki I., Fujimaru R., Suzuki H., Shinomiya N., Takada T., Matsuo H. Identification of a dysfunctional splicing mutation in the SLC22A12/URAT1 gene causing renal hypouricaemia type 1: a report on two families. *Rheumatology (Oxford)* 2020; 59(12): 3988–3990. <doi:10.1093/rheumatology/keaa461>.

<https://www.ncbi.nlm.nih.gov/pubmed/33011794>

Ko S., Komuro J., Katsumata Y., Shiraishi Y., Kawakami T., Yamada Y., Yuasa S., Kohno T., Kosaki K., Fukuda K. Peripheral pulmonary stenosis with Noonan syndrome treated by balloon pulmonary angioplasty. *Pulm Circ* 2020; 10(4): 2045894020954310. <doi:10.1177/2045894020954310>.

<https://www.ncbi.nlm.nih.gov/pubmed/33240484>

Kondo Y., Aoyama K., Suzuki H., Hattori A., Hori I., Ito K., Yoshida A., Koroki M., Ueda K., Kosaki K., Saitoh S. De novo 2q36.3q37.1 deletion encompassing TRIP12 and NPPC yields distinct phenotypes. *Hum Genome Var* 2020; 719. <doi:10.1038/s41439-020-0107-1>.

<https://www.ncbi.nlm.nih.gov/pubmed/32528716>

Kosaki R., Kubota M., Uehara T., Suzuki H., Takenouchi T., Kosaki K. Consecutive medical exome analysis at a tertiary center: Diagnostic and health-economic outcomes. *Am J Med Genet A* 2020; 182(7): 1601–1607. <doi:10.1002/ajmg.a.61589>.

<https://www.ncbi.nlm.nih.gov/pubmed/32369273>

Li L., Fong C. Y., Tay C. G., Tae S. K., Suzuki H., Kosaki K., Thong M. K. Infantile neuroaxonal dystrophy in a pair of Malaysian siblings with progressive cerebellar atrophy: Description of an expanded phenotype with novel PLA2G6 variants. *J Clin Neurosci* 2020; 71289–292. <doi:10.1016/j.jocn.2019.08.111>.

<https://www.ncbi.nlm.nih.gov/pubmed/31493991>

Murakami H., Tsurusaki Y., Enomoto K., Kuroda Y., Yokoi T., Furuya N., Yoshihashi H., Minatogawa M., Abe-Hatano C., Ohashi I., Nishimura N., Kumaki T., Enomoto Y., Naruto T., Iwasaki F., Harada N., Ishikawa A., Kawame H., Sameshima K., Yamaguchi Y., Kobayashi M., Tominaga M., Ishikiriyama S., Tanaka T., Suzumura H., Ninomiya S., Kondo A., Kaname T., Kosaki K., Masuno M., Kuroki Y., Kurosawa K. Update of the genotype and phenotype of KMT2D and KDM6A by genetic screening of 100 patients with clinically suspected Kabuki syndrome. *Am J Med Genet A* 2020; 182(10): 2333–2344. <doi:10.1002/ajmg.a.61793>.

<https://www.ncbi.nlm.nih.gov/pubmed/32803813>

Murakami H., Uehara T., Tsurusaki Y., Enomoto Y., Kuroda Y., Aida N., Kosaki K., Kurosawa K. Blended phenotype of AP4E1 deficiency and Angelman syndrome caused by paternal isodisomy of chromosome 15. *Brain Dev* 2020; 42(3): 289–292.

<doi:10.1016/j.braindev.2019.12.008>.

<https://www.ncbi.nlm.nih.gov/pubmed/31955925>

Oiso N., Kubo A., Shimizu A., Suzuki H., Kosaki K., Chikugo T., Nakabayashi K., Hata K., Yanagihara S., Ishikawa O., Matsubara Y., Amagai M., Kawada A. Epidermodysplasia verruciformis without progression to squamous cell carcinomas in an elderly man: alpha-human papillomavirus infection in the evolving verruca. *Int J Dermatol* 2020; 59(9): e334–e336. <doi:10.1111/ijd.14883>.

<https://www.ncbi.nlm.nih.gov/pubmed/32406058>

Saettini F., Herriot R., Prada E., Nizon M., Zama D., Marzollo A., Romaniouk I., Lougaris V., Cortesi M., Morreale A., Kosaki R., Cardinale F., Ricci S., Dominguez-Garrido E., Montin D., Vincent M., Milani D., Biondi A., Gervasini C., Badolato R. Prevalence of

Immunological Defects in a Cohort of 97 Rubinstein–Taybi Syndrome Patients. *J Clin Immunol* 2020; 40(6): 851–860. <doi:10.1007/s10875-020-00808-4>.

<https://www.ncbi.nlm.nih.gov/pubmed/32594341>

Sakaguchi Y., Uehara T., Sasaki M., Fujimura K., Kishi K., Kosaki K., Takenouchi T. Hereditary spastic paraplegia masqueraded by congenital melanocytic nevus syndrome: Dual pathogenesis of germline non-mosaicism and somatic mosaicism. *Eur J Med Genet* 2020; 63(4): 103803. <doi:10.1016/j.ejmg.2019.103803>.

<https://www.ncbi.nlm.nih.gov/pubmed/31698101>

Sunaga Y., Muramatsu K., Kosaki K., Sugai K., Mizuno T., Kouno M., Tashiro M. Variant in the neuronal vesicular SNARE VAMP2 (synaptobrevin-2): First report in Japan. *Brain Dev* 2020; 42(7): 529–533. <doi:10.1016/j.braindev.2020.04.001>.

<https://www.ncbi.nlm.nih.gov/pubmed/32336483>

Suzuki H., Yamada M., Uehara T., Takenouchi T., Kosaki K. Parallel detection of single nucleotide variants and copy number variants with exome analysis: Validation in a cohort of 700 undiagnosed patients. *Am J Med Genet A* 2020; 182(11): 2529–2532. <doi:10.1002/ajmg.a.61822>.

<https://www.ncbi.nlm.nih.gov/pubmed/32779332>

Suzuki–Muromoto S., Kosaki R., Kosaki K., Kubota M. Familial hemiplegic migraine with a PRRT2 mutation: Phenotypic variations and carbamazepine efficacy. *Brain Dev* 2020; 42(3): 293–297. <doi:10.1016/j.braindev.2019.12.007>.

<https://www.ncbi.nlm.nih.gov/pubmed/31902651>

Takenouchi T., Yamada T., Kashiwagi Y., Yamaguchi Y., Uehara T., Kosaki K. Hypercoagulopathy Associated With Uniparental Disomy of Chromosome 2. *J Pediatr Hematol Oncol* 2020; 42(5): 370–371. <doi:10.1097/MPH.0000000000001834>.

<https://www.ncbi.nlm.nih.gov/pubmed/32487849>

Takeshita Y., Ohto T., Enokizono T., Tanaka M., Suzuki H., Fukushima H., Uehara T., Takenouchi T., Kosaki K., Takada H. Novel ARX mutation identified in infantile spasm syndrome patient. *Hum Genome Var* 2020; 79. <doi:10.1038/s41439-020-0094-2>.

<https://www.ncbi.nlm.nih.gov/pubmed/32257294>

Taruscio D., Baynam G., Cederroth H., Groft S. C., Klee E. W., Kosaki K., Lasko P., Melegh B., Riess O., Salvatore M., Gahl W. A. The Undiagnosed Diseases Network International: Five years and more! *Mol Genet Metab* 2020; 129(4): 243–254.

<doi:10.1016/j.ymgme.2020.01.004>.

<https://www.ncbi.nlm.nih.gov/pubmed/32033911>

Uehara T., Abe K., Oginuma M., Ishitani S., Yoshihashi H., Okamoto N., Takenouchi T., Kosaki K., Ishitani T. Pathogenesis of CDK8-associated disorder: two patients with novel CDK8 variants and in vitro and in vivo functional analyses of the variants. *Sci Rep* 2020; 10(1): 17575. <doi:10.1038/s41598-020-74642-4>.

<https://www.ncbi.nlm.nih.gov/pubmed/33067521>

Uehara T., Yamada M., Umetsu S., Nittono H., Suzuki H., Fujisawa T., Takenouchi T., Inui A., Kosaki K. Biallelic Mutations in the LSR Gene Cause a Novel Type of Infantile Intrahepatic Cholestasis. *J Pediatr* 2020; 221:251–254. <doi:10.1016/j.jpeds.2020.01.064>.

<https://www.ncbi.nlm.nih.gov/pubmed/32303357>

Yamada M., Shiraishi Y., Uehara T., Suzuki H., Takenouchi T., Abe-Hatano C., Kurosawa K., Kosaki K. Diagnostic utility of integrated analysis of exome and transcriptome: Successful diagnosis of Au-Kline syndrome in a patient with submucous cleft palate, scaphocephaly, and intellectual disabilities. *Mol Genet Genomic Med* 2020; 8(9): e1364.

<doi:10.1002/mgg3.1364>.

<https://www.ncbi.nlm.nih.gov/pubmed/32588992>

Yamada M., Sokoda T., Uehara T., Suzuki H., Takenouchi T., Yagihashi T., Maruo Y., Kosaki K. Learning disability and myoclonic epilepsy associated with apparently synonymous but splice-disrupting JMJD1C variant that led to 21 bp deletion of the transcript. *Am J Med Genet A* 2020; 182(12): 3064–3067. <doi:10.1002/ajmg.a.61892>.

<https://www.ncbi.nlm.nih.gov/pubmed/32996679>

Yamada M., Uehara T., Suzuki H., Takenouchi T., Inui A., Ikemiyagi M., Kamimaki I., Kosaki K. Shortfall of exome analysis for diagnosis of Shwachman-Diamond syndrome: Mismapping due to the pseudogene SBDSP1. *Am J Med Genet A* 2020; 182(7): 1631–1636. <doi:10.1002/ajmg.a.61598>.

<https://www.ncbi.nlm.nih.gov/pubmed/32412173>

Yamada M., Uehara T., Suzuki H., Takenouchi T., Kosaki K. Protein elongation variant of PUF60: Milder phenotypic end of the Verheij syndrome. *Am J Med Genet A* 2020; 182(11): 2709–2714. <doi:10.1002/ajmg.a.61816>.

<https://www.ncbi.nlm.nih.gov/pubmed/32851780>

Yamaguchi H., Morisada N., Maruyama A., Kosaki K., Nomura K. Improvement of opsoclonus after congenital cataract surgery in an infant. *Pediatr Int* 2020; 62(1): 108–109. <doi:10.1111/ped.14039>.

<https://www.ncbi.nlm.nih.gov/pubmed/31957102>

Yokoi T., Enomoto Y., Uehara T., Kosaki K., Kurosawa K. A Japanese girl with mild xeroderma pigmentosum group D neurological disease diagnosed using whole-exome sequencing. *Hum Genome Var* 2020; 722. <doi:10.1038/s41439-020-0109-z>.

<https://www.ncbi.nlm.nih.gov/pubmed/32802388>

<栄養障害、代謝性疾患、消化器疾患>

Mizuno K., Shimizu T., Ida S., Ito S., Inokuchi M., Ohura T., Okumura A., Kawai M., Kikuchi T., Sakurai M., Sugihara S., Suzuki M., Takitani K., Tanaka D., Mushiake S., Yoshiike N., Kodama H., Okada K., Tsutsumi C., Hara M., Hanawa Y., Kawakami K., Inomata H., Oguni T., Bito Y., Uchida K., Sugiyama A. Policy statement of enteral nutrition for preterm and very low birthweight infants. *Pediatr Int* 2020; 62(2): 124–127. <doi:10.1111/ped.14067>.

<https://www.ncbi.nlm.nih.gov/pubmed/32026585>

Uehara T., Yamada M., Umetsu S., Nittono H., Suzuki H., Fujisawa T., Takenouchi T., Inui A., Kosaki K. Biallelic Mutations in the LSR Gene Cause a Novel Type of Infantile Intrahepatic Cholestasis. *J Pediatr* 2020; 221251–254. <doi:10.1016/j.jpeds.2020.01.064>.

<https://www.ncbi.nlm.nih.gov/pubmed/32303357>

<血液疾患、腫瘍>

Aoki T., Kishimoto H., Hirato J., Kanemura Y., Mori M., Arakawa Y., Kurihara J., Oguma E., Koh K. A 21-Month-Old Boy with One-Month History of Vomiting. *Brain Pathol* 2020; 30(2): 425–426. <doi:10.1111/bpa.12823>.

<https://www.ncbi.nlm.nih.gov/pubmed/32100428>

Carcao M., Kearney S., Lu M. Y., Taki M., Rubens D., Shen C., Santagostino E. Long-Term

Safety and Efficacy of Nonacog Beta Pegol (N9-GP) Administered for at Least 5 Years in Previously Treated Children with Hemophilia B. *Thromb Haemost* 2020; 120(5): 737–746. <doi:10.1055/s-0040-1709521>.

<https://www.ncbi.nlm.nih.gov/pubmed/32369845>

Dingler F. A., Wang M., Mu A., Millington C. L., Oberbeck N., Watcham S., Pontel L. B., Kamimae-Lanning A. N., Langevin F., Nadler C., Cordell R. L., Monks P. S., Yu R., Wilson N. K., Hira A., Yoshida K., Mori M., Okamoto Y., Okuno Y., Muramatsu H., Shiraishi Y., Kobayashi M., Moriguchi T., Osumi T., Kato M., Miyano S., Ito E., Kojima S., Yabe H., Yabe M., Matsuo K., Ogawa S., Gottgens B., Hodskinson M. R. G., Takata M., Patel K. J. Two Aldehyde Clearance Systems Are Essential to Prevent Lethal Formaldehyde Accumulation in Mice and Humans. *Mol Cell* 2020; 80(6): 996–1012 e1019. <doi:10.1016/j.molcel.2020.10.012>.

<https://www.ncbi.nlm.nih.gov/pubmed/33147438>

Hara Y., Shiba N., Yamato G., Ohki K., Tabuchi K., Sotomatsu M., Tomizawa D., Kinoshita A., Arakawa H., Saito A. M., Kiyokawa N., Tawa A., Horibe K., Taga T., Adachi S., Taki T., Hayashi Y. Patients aged less than 3 years with acute myeloid leukaemia characterize a molecularly and clinically distinct subgroup. *Br J Haematol* 2020; 188(4): 528–539. <doi:10.1111/bjh.16203>.

<https://www.ncbi.nlm.nih.gov/pubmed/31612466>

Hasegawa D., Tawa A., Tomizawa D., Watanabe T., Saito A. M., Kudo K., Taga T., Iwamoto S., Shimada A., Terui K., Moritake H., Kinoshita A., Takahashi H., Nakayama H., Koh K., Goto H., Kosaka Y., Miyachi H., Horibe K., Nakahata T., Adachi S. Attempts to optimize postinduction treatment in childhood acute myeloid leukemia without core-binding factors: A report from the Japanese Pediatric Leukemia/Lymphoma Study Group (JPLSG). *Pediatr Blood Cancer* 2020; 67(12): e28692. <doi:10.1002/pbc.28692>.

<https://www.ncbi.nlm.nih.gov/pubmed/32886449>

Hiyama E., Hishiki T., Watanabe K., Ida K., Ueda Y., Kurihara S., Yano M., Hoshino K., Yokoi A., Takama Y., Nogami Y., Taguchi T., Mori M., Kihira K., Miyazaki O., Fuji H., Honda S., Iehara T., Kazama T., Fujimura J., Tanaka Y., Inoue T., Tajiri T., Kondo S., Oue T., Yoshimura K. Outcome and Late Complications of Hepatoblastomas Treated Using the Japanese Study Group for Pediatric Liver Tumor 2 Protocol. *J Clin Oncol* 2020; 38(22): 2488–2498. <doi:10.1200/JCO.19.01067>.

<https://www.ncbi.nlm.nih.gov/pubmed/32421442>

Inoue A., Imamura C. K., Shimada H., Katayama D., Urabe K., Suzuki R., Takitani K., Ashida A. Pharmacokinetics, Efficacy and Safety of Bosutinib in a Pediatric Patient With Chronic Myeloid Leukemia. *J Pediatr Pharmacol Ther* 2020; 25(8): 742-745. <doi:10.5863/1551-6776-25.8.742>.

<https://www.ncbi.nlm.nih.gov/pubmed/33214787>

Isshiki K., Shima H., Yamazaki F., Takenouchi T., Shimada H. A Case of Pulmonary Venocclusive Disease Following Hepatic Venocclusive Disease After Autologous Hematopoietic Stem Cell Transplantation for Neuroblastoma. *J Pediatr Hematol Oncol* 2020; 42(7): e677-e679. <doi:10.1097/MPH.0000000000001566>.

<https://www.ncbi.nlm.nih.gov/pubmed/31335821>

Ito J., Nakano Y., Shima H., Miwa T., Kogure Y., Isshiki K., Yamazaki F., Oishi Y., Morimoto Y., Kataoka K., Okita H., Hirato J., Ichimura K., Shimada H. Central nervous system ganglioneuroblastoma harboring MYO5A-NTRK3 fusion. *Brain Tumor Pathol* 2020; 37(3): 105-110. <doi:10.1007/s10014-020-00371-1>.

<https://www.ncbi.nlm.nih.gov/pubmed/32556925>

Kada A., Fukano R., Mori T., Kamei M., Tanaka F., Ueyama J., Sekimizu M., Osumi T., Mori T., Koga Y., Ohki K., Fujita N., Mitsui T., Saito A. M., Hashimoto H., Kobayashi R. A Multicenter, Open-label, Clinical Trial to Assess the Effectiveness and Safety of Allogeneic Hematopoietic Stem Cell Transplantation Using Reduced-intensity Conditioning in Relapsed/refractory Anaplastic Large-cell Lymphoma in Children. *Acta Med Okayama* 2020; 74(1): 89-94. <doi:10.18926/AMO/57959>.

<https://www.ncbi.nlm.nih.gov/pubmed/32099255>

Kim Y., Sudo A., Oyama R., Keino D., Tomizawa D., Kato M., Osumi T., Mori T. Isolated Central Nervous System Progression During Systemic Treatment With Brentuximab Vedotin Monotherapy in a Pediatric Patient With Recurrent ALK-negative Anaplastic Large Cell Lymphoma. *J Pediatr Hematol Oncol* 2020. <doi:10.1097/MPH.0000000000001914>.

<https://www.ncbi.nlm.nih.gov/pubmed/32769561>

Kudo K., Maeda M., Suzuki N., Kanegane H., Ohga S., Ishii E., Shioda Y., Imamura T., Imashuku S., Tsunematsu Y., Endo M., Shimada A., Koga Y., Hashii Y., Noguchi M., Inoue

M., Tabuchi K., Morimoto A., Histiocytosis study group of the Japanese Society of Pediatric Hematology Oncology. Nationwide retrospective review of hematopoietic stem cell transplantation in children with refractory Langerhans cell histiocytosis. *Int J Hematol* 2020; 111(1): 137–148. <doi:10.1007/s12185-019-02760-5>.

<https://www.ncbi.nlm.nih.gov/pubmed/31758416>

Matsuoka Y. J., Okubo R., Shimizu Y., Tsuji K., Narisawa T., Sasaki J., Sasai H., Akashi-Tanaka S., Hamaguchi T., Iwasa T., Iwata S., Kato T., Kurotani K., Maruyama D., Mori A., Ogawa A., Sakurai N., Shimazu T., Shimizu C., Tabuchi T., Takahashi M., Takano T., Tatematsu N., Uchitomi Y., Watanabe C., Fukui T. Developing the structure of Japan's cancer survivorship guidelines using an expert panel and modified Delphi method. *J Cancer Surviv* 2020; 14(3): 273–283. <doi:10.1007/s11764-019-00840-3>.

<https://www.ncbi.nlm.nih.gov/pubmed/31811478>

Mitsui T., Fujita N., Koga Y., Fukano R., Osumi T., Hama A., Koh K., Kakuda H., Inoue M., Fukuda T., Yabe H., Takita J., Shimada A., Hashii Y., Sato A., Atsuta Y., Kanda Y., Suzumiya J., Kobayashi R. The effect of graft-versus-host disease on outcomes after allogeneic stem cell transplantation for refractory lymphoblastic lymphoma in children and young adults. *Pediatr Blood Cancer* 2020; 67(4): e28129. <doi:10.1002/pbc.28129>.

<https://www.ncbi.nlm.nih.gov/pubmed/31876367>

Mori M., Hira A., Yoshida K., Muramatsu H., Okuno Y., Shiraishi Y., Anmae M., Yasuda J., Tadaka S., Kinoshita K., Osumi T., Noguchi Y., Adachi S., Kobayashi R., Kawabata H., Imai K., Morio T., Tamura K., Takaori-Kondo A., Yamamoto M., Miyano S., Kojima S., Ito E., Ogawa S., Matsuo K., Yabe H., Yabe M., Takata M. Pathogenic mutations identified by a multimodality approach in 117 Japanese Fanconi anemia patients. *Haematologica* 2020; 105(4): 1166–1167. <doi:10.3324/haematol.2019.245720>.

<https://www.ncbi.nlm.nih.gov/pubmed/32238468>

Ohki K., Takahashi H., Fukushima T., Nanmoku T., Kusano S., Mori M., Nakazawa Y., Yuza Y., Migita M., Okuno H., Morimoto A., Yoshino H., Kato M., Hayashi Y., Manabe A., Ohara A., Hasegawa D., Inukai T., Tomizawa D., Koh K., Kiyokawa N., Tokyo Children's Cancer Study Group. Impact of immunophenotypic characteristics on genetic subgrouping in childhood acute lymphoblastic leukemia: Tokyo Children's Cancer Study Group (TCCSG) study L04-16. *Genes Chromosomes Cancer* 2020; 59(10): 551–561. <doi:10.1002/gcc.22858>.

<https://www.ncbi.nlm.nih.gov/pubmed/32368831>

Osumi T., Yoshimura S., Sako M., Uchiyama T., Ishikawa T., Kawai T., Inoue E., Takimoto T., Takeuchi I., Yamada M., Sakamoto K., Yoshida K., Kimura Y., Matsukawa Y., Matsumoto K., Imadome K. I., Arai K., Deguchi T., Imai K., Yuza Y., Matsumoto K., Onodera M., Kanegane H., Tomizawa D., Kato M. Prospective Study of Allogeneic Hematopoietic Stem Cell Transplantation with Post-Transplantation Cyclophosphamide and Antithymocyte Globulin from HLA-Mismatched Related Donors for Nonmalignant Diseases. *Biol Blood Marrow Transplant* 2020; 26(11): e286–e291. <doi:10.1016/j.bbmt.2020.08.008>.

<https://www.ncbi.nlm.nih.gov/pubmed/32798657>

Sakamoto A., Yamada M., Tsujimoto S. I., Osumi T., Arai K., Tomizawa D., Ishiguro A., Matsumoto K., Imadome K. I., Kato M. A case of human herpesvirus 6 encephalitis following pediatric hematopoietic stem cell transplantation: early diagnosis and treatment matters. *Int J Hematol* 2020; 112(5): 751–754. <doi:10.1007/s12185-020-02905-x>.

<https://www.ncbi.nlm.nih.gov/pubmed/32529583>

Sakamoto K., Osumi T., Yoshimura S., Shimizu S., Kato M., Tomizawa D., Fukuda A., Sakamoto S., Nakano N., Yoshioka T., Miyazaki O., Nosaka S., Deguchi T., Kiyokawa N., Kasahara M., Matsumoto K. Living-donor liver transplantation providing an adequate chemotherapy for a pediatric patient with anaplastic large cell lymphoma complicated with liver failure due to the aggravation of biliary hepatopathy by secondary hemophagocytic lymphohistiocytosis. *Int J Hematol* 2020; 112(6): 900–905. <doi:10.1007/s12185-020-02949-z>.

<https://www.ncbi.nlm.nih.gov/pubmed/32710432>

Shirai R., Osumi T., Terashima K., Kiyotani C., Uchiyama M., Tsujimoto S., Yoshida M., Yoshida K., Uchiyama T., Tomizawa D., Shioda Y., Sekiguchi M., Watanabe K., Keino D., Ueno-Yokohata H., Ohki K., Takita J., Ito S., Deguchi T., Kiyokawa N., Ogiwara H., Hishiki T., Ogawa S., Okita H., Matsumoto K., Yoshioka T., Kato M. High prevalence of SMARCB1 constitutional abnormalities including mosaicism in malignant rhabdoid tumors. *Eur J Hum Genet* 2020; 28(8): 1124–1128. <doi:10.1038/s41431-020-0614-z>.

<https://www.ncbi.nlm.nih.gov/pubmed/32218533>

Takenouchi T., Yamada T., Kashiwagi Y., Yamaguchi Y., Uehara T., Kosaki K. Hypercoagulopathy Associated With Uniparental Disomy of Chromosome 2. *J Pediatr*

Hematol Oncol 2020; 42(5): 370–371. <doi:10.1097/MPH.0000000000001834>.

<https://www.ncbi.nlm.nih.gov/pubmed/32487849>

Tsujimoto S. I., Shirai R., Utano T., Osumi T., Matsumoto K., Shioda Y., Kiyotani C., Uchiyama T., Deguchi T., Terashima K., Tomizawa D., Matsumoto K., Kato M. Comparison of clonazepam and levetiracetam in children for prevention of busulfan-induced seizure in hematopoietic stem cell transplantation. *Int J Hematol* 2020; 111(3): 463–466.

<doi:10.1007/s12185-019-02795-8>.

<https://www.ncbi.nlm.nih.gov/pubmed/31863341>

Tsumura Y., Yamada Y., Osumi T., Kato M., Terashima K., Shioda Y., Kiyotani C., Matsumoto K., Tomizawa D. Successful Treatment With ATRA and Arsenic Trioxide for a Child With Down Syndrome and Acute Promyelocytic Leukemia. *J Pediatr Hematol Oncol* 2020; 42(4): 322–325. <doi:10.1097/MPH.0000000000001438>.

<https://www.ncbi.nlm.nih.gov/pubmed/30807394>

Utano T., Kato M., Osumi T., Shioda Y., Kiyotani C., Terashima K., Tomizawa D., Matsumoto K., Yamatani A. Tacrolimus blood concentration increase depends on administration route when combined with voriconazole in pediatric stem cell transplant recipients. *Pediatr Transplant* 2020; 24(1): e13619. <doi:10.1111/petr.13619>.

<https://www.ncbi.nlm.nih.gov/pubmed/31820535>

Yamanaka J., Shimizu M., Sato M., Inoue M., Matsui M., Shimada H., Shichino H. A Case of Childhood Blastic Phase Chronic Myeloid Leukemia With Minor BCR-ABL. *J Pediatr Hematol Oncol* 2020; 42(6): e459–e462. <doi:10.1097/MPH.0000000000001488>.

<https://www.ncbi.nlm.nih.gov/pubmed/30994506>

Yoshida M., Tanase-Nakao K., Shima H., Shirai R., Yoshida K., Osumi T., Deguchi T., Mori M., Arakawa Y., Takagi M., Miyamura T., Sakaguchi K., Toyoda H., Ishida H., Sakata N., Imamura T., Kawahara Y., Morimoto A., Koike T., Yagasaki H., Ito S., Tomizawa D., Kiyokawa N., Narumi S., Kato M. Prevalence of germline GATA2 and SAMD9/9L variants in paediatric haematological disorders with monosomy 7. *Br J Haematol* 2020; 191(5): 835–843. <doi:10.1111/bjh.17006>.

<https://www.ncbi.nlm.nih.gov/pubmed/32770553>

Yoshida N., Sakaguchi H., Yabe M., Hasegawa D., Hama A., Hasegawa D., Kato M.,

Noguchi M., Terui K., Takahashi Y., Cho Y., Sato M., Koh K., Kakuda H., Shimada H., Hashii Y., Sato A., Kato K., Atsuta Y., Watanabe K., Pediatric Myelodysplastic Syndrome Working Group of the Japan Society for Hematopoietic Cell Transplantation. Clinical Outcomes after Allogeneic Hematopoietic Stem Cell Transplantation in Children with Juvenile Myelomonocytic Leukemia: A Report from the Japan Society for Hematopoietic Cell Transplantation. *Biol Blood Marrow Transplant* 2020; 26(5): 902–910.

<doi:10.1016/j.bbmt.2019.11.029>.

<https://www.ncbi.nlm.nih.gov/pubmed/31790827>

<呼吸器疾患、アレルギー>

Adachi T., Kainuma K., Asano K., Amagai M., Arai H., Ishii K. J., Ito K., Uchio E., Ebisawa M., Okano M., Kabashima K., Kondo K., Konno S., Saeki H., Sonobe M., Nagao M., Hizawa N., Fukushima A., Fujieda S., Matsumoto K., Morita H., Yamamoto K., Yoshimoto A., Tamari M. Strategic Outlook toward 2030: Japan's research for allergy and immunology – Secondary publication. *Allergol Int* 2020; 69(4): 561–570. <doi:10.1016/j.alit.2020.04.006>.

<https://www.ncbi.nlm.nih.gov/pubmed/32600925>

Fukushima N., Shimojima N., Ishitate M., Miyakawa T., Hirobe S., Miura M. Clinical and structural aspects of tracheal stenosis and a novel embryological hypothesis of left pulmonary artery sling. *Pediatr Pulmonol* 2020; 55(3): 747–753. <doi:10.1002/ppul.24661>.

<https://www.ncbi.nlm.nih.gov/pubmed/31975532>

Kobayashi H., Takimoto T., Kitaoka H., Kijima T. Aerosol spread with use of high-flow nasal cannulae: a computational fluid dynamics analysis. *J Hosp Infect* 2020; 106(1): 204–205.

<doi:10.1016/j.jhin.2020.06.010>.

<https://www.ncbi.nlm.nih.gov/pubmed/32544507>

Matsumoto K., Iikura K., Morita H., Saito H. Barrier dysfunction in the atopic march—how does atopic dermatitis lead to asthma in children? *J Allergy Clin Immunol* 2020; 145(6): 1551–1553. <doi:10.1016/j.jaci.2020.04.014>.

<https://www.ncbi.nlm.nih.gov/pubmed/32344057>

Ohno M., Fuchimoto Y., Higuchi M., Yamaoka T., Komura M., Umezawa A., Hsu H. C., Enosawa S., Kuroda T. Long-term observation of airway reconstruction using decellularized tracheal allografts in micro-miniature pigs at growing stage. *Regen Ther* 2020; 1564–69.

<doi:10.1016/j.reth.2020.04.010>.

<https://www.ncbi.nlm.nih.gov/pubmed/33426203>

Takahashi N., Fuchimoto Y., Mori T., Abe K., Yamada Y., Koinuma G., Kuroda T. Post-esophageal atresia repair double acquired tracheoesophageal fistulas treated successfully by gastric transposition: a case report. *Surg Case Rep* 2020; 6(1): 224. <doi:10.1186/s40792-020-01004-7>.

<https://www.ncbi.nlm.nih.gov/pubmed/32975613>

Unno H., Arae K., Matsuda A., Ikutani M., Tamari M., Motomura K., Toyama S., Suto H., Okumura K., Matsuda A., Morita H., Sudo K., Saito H., Matsumoto K., Nakae S. Critical role of IL-33, but not IL-25 or TSLP, in silica crystal-mediated exacerbation of allergic airway eosinophilia. *Biochem Biophys Res Commun* 2020; 533(3): 493-500. <doi:10.1016/j.bbrc.2020.09.046>.

<https://www.ncbi.nlm.nih.gov/pubmed/32977946>

<循環器疾患>

Fujisawa T., Aizawa Y., Katsumata Y., Kimura K., Hashimoto K., Yamashita T., Miyama H., Kimura T., Kosaki K., Takatsuki S., Shimizu W., Fukuda K. Mexiletine shortens the QT interval in a pedigree of KCNH2 related long QT syndrome. *J Arrhythm* 2020; 36(1): 193-196. <doi:10.1002/joa3.12300>.

<https://www.ncbi.nlm.nih.gov/pubmed/32071644>

Fukushima N., Shimojima N., Ishitate M., Miyakawa T., Hirobe S., Miura M. Clinical and structural aspects of tracheal stenosis and a novel embryological hypothesis of left pulmonary artery sling. *Pediatr Pulmonol* 2020; 55(3): 747-753. <doi:10.1002/ppul.24661>.

<https://www.ncbi.nlm.nih.gov/pubmed/31975532>

Iio K., Fukushima N., Akamine K., Uda K., Hataya H., Miura M. Acute Rheumatic Fever and Kawasaki Disease Occurring in a Single Patient. *Front Pediatr* 2020; 8562. <doi:10.3389/fped.2020.00562>.

<https://www.ncbi.nlm.nih.gov/pubmed/33014943>

Ishizaki-Asami R., Uchida K., Tsuchihashi T., Shibata A., Kodo K., Emoto K., Mikoshiba K., Takahashi T., Yamagishi H. Inositol 1,4,5-trisphosphate receptor 2 as a novel marker of vasculature to delineate processes of cardiopulmonary development. *Dev Biol* 2020; 458(2): 237-245. <doi:10.1016/j.ydbio.2019.11.011>.

<https://www.ncbi.nlm.nih.gov/pubmed/31758944>

Sumitomo N. F., Fukushima N., Miura M. Flecainide improves cardiac synchronization in an early infant with Wolff–Parkinson–White syndrome with left ventricular dyssynchrony. *J Cardiol Cases* 2020; 22(1): 1–4. <doi:10.1016/j.jccase.2020.03.004>.

<https://www.ncbi.nlm.nih.gov/pubmed/32636959>

<新生児疾患>

Arimitsu T., Wakabayashi D., Tamaoka S., Takahashi M., Hida M., Takahashi T. Case Report: Intact Survival of a Marginally Viable Male Infant Born Weighing 268 Grams at 24 Weeks Gestation. *Front Pediatr* 2020; 8628362. <doi:10.3389/fped.2020.628362>.

<https://www.ncbi.nlm.nih.gov/pubmed/33614546>

<神経・筋疾患、心身症>

Ikenori M., Yotani N., Yamada M., Imadome K. I., Miyairi I., Ishiguro A. Eleven-Year-Old Girl with Acute Genital Ulcers: Was It Sexual Abuse? *J Paediatr Child Health* 2020; 56(12): 1995–1996. <doi:10.1111/jpc.15210>.

<https://www.ncbi.nlm.nih.gov/pubmed/33351258>

Kamidani S., Shoji K., Ogawa E., Funaki T., Mishina H., Miyairi I. High Rate of Febrile Seizures in Japanese Children With Occult Bacteremia. *Pediatr Emerg Care* 2020; 36(4): e199–e203. <doi:10.1097/PEC.0000000000001274>.

<https://www.ncbi.nlm.nih.gov/pubmed/28953097>

Kobayashi Y., Takeda T., Kunitomi H., Ueki A., Misu K., Kowashi A., Takahashi T., Anko M., Watanabe K., Masuda K., Uchida T., Tominaga E., Banno K., Kosaki K., Aoki D. Cowden syndrome complicated by schizophrenia: A first clinical report. *Eur J Med Genet* 2020; 63(8): 103959. <doi:10.1016/j.ejmg.2020.103959>.

<https://www.ncbi.nlm.nih.gov/pubmed/32461083>

Muller P. A., Schneeberger M., Matheis F., Wang P., Kerner Z., Ilanges A., Pellegrino K., Del Marmol J., Castro T. B. R., Furuichi M., Perkins M., Han W., Rao A., Pickard A. J., Cross J. R., Honda K., de Araujo I., Mucida D. Microbiota modulate sympathetic neurons via a gut–brain circuit. *Nature* 2020; 583(7816): 441–446. <doi:10.1038/s41586-020-2474-7>.

<https://www.ncbi.nlm.nih.gov/pubmed/32641826>

Nishiya K., Sekiguchi S., Yoshimura H., Takamura A., Wada H., Konishi E., Saiki T., Tsunekawa K., Fujisaki K., Suzuki Y. Good clinical teachers in pediatrics: The perspective of pediatricians in Japan. *Pediatr Int* 2020; 62(5): 549–555. <doi:10.1111/ped.14125>.

<https://www.ncbi.nlm.nih.gov/pubmed/31883414>

Sakaguchi Y., Uehara T., Sasaki M., Fujimura K., Kishi K., Kosaki K., Takenouchi T. Hereditary spastic paraplegia masqueraded by congenital melanocytic nevus syndrome: Dual pathogenesis of germline non-mosaicism and somatic mosaicism. *Eur J Med Genet* 2020; 63(4): 103803. <doi:10.1016/j.ejmg.2019.103803>.

<https://www.ncbi.nlm.nih.gov/pubmed/31698101>

Sasaki Y., Yagihashi T., Kasahara M., Usami M., Kono T., Okada T. Clinical implications of a history of stealing on psychiatric disorders in children and adolescents. *PLoS One* 2020; 15(8): e0237906. <doi:10.1371/journal.pone.0237906>.

<https://www.ncbi.nlm.nih.gov/pubmed/32853286>

Takenouchi T., Yamada T., Kashiwagi Y., Yamaguchi Y., Uehara T., Kosaki K. Hypercoagulopathy Associated With Uniparental Disomy of Chromosome 2. *J Pediatr Hematol Oncol* 2020; 42(5): 370–371. <doi:10.1097/MPH.0000000000001834>.

<https://www.ncbi.nlm.nih.gov/pubmed/32487849>

Takeshita Y., Ohto T., Enokizono T., Tanaka M., Suzuki H., Fukushima H., Uehara T., Takenouchi T., Kosaki K., Takada H. Novel ARX mutation identified in infantile spasm syndrome patient. *Hum Genome Var* 2020; 79. <doi:10.1038/s41439-020-0094-2>.

<https://www.ncbi.nlm.nih.gov/pubmed/32257294>

Yamada M., Shiraishi Y., Uehara T., Suzuki H., Takenouchi T., Abe-Hatano C., Kurosawa K., Kosaki K. Diagnostic utility of integrated analysis of exome and transcriptome: Successful diagnosis of Au-Kline syndrome in a patient with submucous cleft palate, scaphocephaly, and intellectual disabilities. *Mol Genet Genomic Med* 2020; 8(9): e1364.

<doi:10.1002/mgg3.1364>.

<https://www.ncbi.nlm.nih.gov/pubmed/32588992>

Yamada M., Sokoda T., Uehara T., Suzuki H., Takenouchi T., Yagihashi T., Maruo Y., Kosaki K. Learning disability and myoclonic epilepsy associated with apparently synonymous but splice-disrupting JMJD1C variant that led to 21 bp deletion of the transcript. *Am J Med*

Genet A 2020; 182(12): 3064–3067. <doi:10.1002/ajmg.a.61892>.

<https://www.ncbi.nlm.nih.gov/pubmed/32996679>

Yamashita Y., Ogawa T., Ogaki K., Kamo H., Sukigara T., Kitahara E., Izawa N., Iwamuro H., Oyama G., Kamagata K., Hatano T., Umemura A., Kosaki R., Kubota M., Shimo Y., Hattori N. Neuroimaging evaluation and successful treatment by using directional deep brain stimulation and levodopa in a patient with GNAO1-associated movement disorder: A case report. *J Neurol Sci* 2020; 411116710. <doi:10.1016/j.jns.2020.116710>.

<https://www.ncbi.nlm.nih.gov/pubmed/32044685>

<腎・泌尿器疾患、生殖器疾患>

Aoki Y., Hamasaki Y., Satoh H., Matsui Z., Muramatsu M., Hamada R., Harada R., Ishikura K., Hataya H., Honda M., Sakai K., Shishido S. Long-term outcomes of pediatric kidney transplantation: A single-center experience over the past 34 years in Japan. *Int J Urol* 2020; 27(2): 172–178. <doi:10.1111/iju.14160>.

<https://www.ncbi.nlm.nih.gov/pubmed/31826334>

Awazu M., Hida M. Folic acid supplementation alleviates reduced ureteric branching, nephrogenesis, and global DNA methylation induced by maternal nutrient restriction in rat embryonic kidney. *PLoS One* 2020; 15(4): e0230289. <doi:10.1371/journal.pone.0230289>.

<https://www.ncbi.nlm.nih.gov/pubmed/32251454>

Dote Y., Kibe T., Murakami T., Awazu M. Ask-Upmark kidney in a girl with neurofibromatosis type 1. *CEN Case Rep* 2020; 9(3): 285–288. <doi:10.1007/s13730-020-00470-0>.

<https://www.ncbi.nlm.nih.gov/pubmed/32277359>

Gotoh Y., Shishido S., Hamasaki Y., Watarai Y., Hattori M., Miura K., Ishizuka K., Fujita N., Saito K., Nakagawa Y., Hotta K., Hataya H., Hamada R., Sato H., Kitayama H., Ishikura K., Honda M., Uemura O. Kidney function of Japanese children undergoing kidney transplant with preemptive therapy for cytomegalovirus infection. *Transpl Infect Dis* 2020; 22(3): e13271. <doi:10.1111/tid.13271>.

<https://www.ncbi.nlm.nih.gov/pubmed/32108410>

Hamasaki Y., Hamada R., Muramatsu M., Matsumoto S., Aya K., Ishikura K., Kaneko T., Iijima K. A cross-sectional nationwide survey of congenital and infantile nephrotic syndrome

in Japan. *BMC Nephrol* 2020; 21(1): 363. <doi:10.1186/s12882-020-02010-5>.

<https://www.ncbi.nlm.nih.gov/pubmed/32838745>

Hirano D., Inoue E., Sako M., Ashida A., Honda M., Takahashi S., Iijima K., Hattori M., Japanese Society of Pediatric Nephrology. Clinical characteristics at the renal replacement therapy initiation of Japanese pediatric patients: a nationwide cross-sectional study. *Clin Exp Nephrol* 2020; 24(1): 82-87. <doi:10.1007/s10157-019-01788-5>.

<https://www.ncbi.nlm.nih.gov/pubmed/31541336>

Ito S., Torii T., Nakajima A., Iijima T., Murano H., Horiuchi H., Yamanaka H., Honda M. Prevalence of gout and asymptomatic hyperuricemia in the pediatric population: a cross-sectional study of a Japanese health insurance database. *BMC Pediatr* 2020; 20(1): 481. <doi:10.1186/s12887-020-02379-0>.

<https://www.ncbi.nlm.nih.gov/pubmed/33059648>

Jia X., Yamamura T., Gbadegesin R., McNulty M. T., Song K., Nagano C., Hitomi Y., Lee D., Aiba Y., Khor S. S., Ueno K., Kawai Y., Nagasaki M., Noiri E., Horinouchi T., Kaito H., Hamada R., Okamoto T., Kamei K., Kaku Y., Fujimaru R., Tanaka R., Shima Y., Research Consortium on Genetics of Childhood Idiopathic Nephrotic Syndrome in Japan, Baek J., Kang H. G., Ha I. S., Han K. H., Yang E. M., Korean Consortium of Hereditary Renal Diseases in Children, Abeyagunawardena A., Lane B., Chryst-Stangl M., Esezobor C., Solarin A., Midwest Pediatric Nephrology Consortium, Dossier C., Deschenes G., Nephrovir, Vivarelli M., Debiec H., Ishikura K., Matsuo M., Nozu K., Ronco P., Cheong H. I., Sampson M. G., Tokunaga K., Iijima K. Common risk variants in NPHS1 and TNFSF15 are associated with childhood steroid-sensitive nephrotic syndrome. *Kidney Int* 2020; 98(5): 1308-1322. <doi:10.1016/j.kint.2020.05.029>.

<https://www.ncbi.nlm.nih.gov/pubmed/32554042>

Kawamura Y., Toyoda Y., Ohnishi T., Hisatomi R., Higashino T., Nakayama A., Shimizu S., Yanagi M., Kamimaki I., Fujimaru R., Suzuki H., Shinomiya N., Takada T., Matsuo H. Identification of a dysfunctional splicing mutation in the SLC22A12/URAT1 gene causing renal hypouricaemia type 1: a report on two families. *Rheumatology (Oxford)* 2020; 59(12): 3988-3990. <doi:10.1093/rheumatology/keaa461>.

<https://www.ncbi.nlm.nih.gov/pubmed/33011794>

Maruyama Y., Sato M., Inaba Y., Fukuyama T. Comparison of mild encephalopathy with

reversible splenic lesion with and without acute focal bacterial nephritis. *Brain Dev* 2020; 42(1): 56–63. <doi:10.1016/j.braindev.2019.08.008>.

<https://www.ncbi.nlm.nih.gov/pubmed/31591022>

Minamikawa S., Miwa S., Inagaki T., Nishiyama K., Kaito H., Ninchoji T., Yamamura T., Nagano C., Sakakibara N., Ishimori S., Hara S., Yoshikawa N., Hirano D., Harada R., Hamada R., Matsunoshita N., Nagata M., Shima Y., Nakanishi K., Nagase H., Takeda H., Morisada N., Iijima K., Nozu K. Molecular mechanisms determining severity in patients with Pierson syndrome. *J Hum Genet* 2020; 65(4): 355–362. <doi:10.1038/s10038-019-0715-0>.

<https://www.ncbi.nlm.nih.gov/pubmed/31959872>

Morisada N., Hamada R., Miura K., Ye M. J., Nozu K., Hattori M., Iijima K. Bardet–Biedl syndrome in two unrelated patients with identical compound heterozygous SCLT1 mutations. *CEN Case Rep* 2020; 9(3): 260–265. <doi:10.1007/s13730-020-00472-y>.

<https://www.ncbi.nlm.nih.gov/pubmed/32253632>

Morizawa Y., Satoh H., Iwasa S., Sato A., Aoki Y., Hamada R. Clinical Accuracy of Average Creatinine and Cystatin–C–Based Estimated GFR in Japanese Living Renal Transplantation Donors. *Transplant Proc* 2020; 52(10): 3017–3022. <doi:10.1016/j.transproceed.2020.06.013>.

<https://www.ncbi.nlm.nih.gov/pubmed/32711850>

Morizawa Y., Satoh H., Iwasa S., Sato A., Aoki Y., Harada R., Hamada R., Hataya H. Increasing bladder capacity and vesicoureteral reflux in pediatric kidney transplant patients. *Int J Urol* 2020; 27(11): 1008–1012. <doi:10.1111/iju.14348>.

<https://www.ncbi.nlm.nih.gov/pubmed/32789949>

Nozu K., Yamamura T., Horinouchi T., Nagano C., Sakakibara N., Ishikura K., Hamada R., Morisada N., Iijima K. Inherited salt–losing tubulopathy: An old condition but a new category of tubulopathy. *Pediatr Int* 2020; 62(4): 428–437. <doi:10.1111/ped.14089>.

<https://www.ncbi.nlm.nih.gov/pubmed/31830341>

Ohnishi T., Asada N., Furuichi M., Sekiguchi S., Awazu M., Hori N., Kamimaki I. A novel screening method for pediatric urinary tract infection using ordinary diapers. *Sci Rep* 2020; 10(1): 19342. <doi:10.1038/s41598-020-76405-7>.

<https://www.ncbi.nlm.nih.gov/pubmed/33168907>

Ohnishi T., Asato S., Maeda A., Nagata M., Uwamino Y., Kamimaki I. Does the patient truly not have congenital anomalies of the kidney and urinary tract? Urinary tract infection caused by *Gardnerella vaginalis*. *Pediatr Int* 2020; 62(8): 1009–1010. <doi:10.1111/ped.14328>. <https://www.ncbi.nlm.nih.gov/pubmed/32851758>

Ohnishi T., Mishima Y., Takizawa S., Tsutsumi K., Amemiya A., Akiyama N., Kanna Y., Asato S., Tomita M., Ikemiyagi M., Shikoro N., Nakazawa M., Kurihara N., Kamimaki I. Clinical Features of Febrile Urinary Tract Infection Caused by Extended-spectrum Beta-lactamase-producing *Escherichia Coli* in Children. *Keio J Med* 2020; 69(2): 43–47. <doi:10.2302/kjm.2019-0005-OA>. <https://www.ncbi.nlm.nih.gov/pubmed/31474677>

Saida K., Kamei K., Hamada R., Yoshikawa T., Kano Y., Nagata H., Sato M., Ogura M., Harada R., Hataya H., Miyazaki O., Nosaka S., Ito S., Ishikura K. A simple, refined approach to diagnosing renovascular hypertension in children: A 10-year study. *Pediatr Int* 2020; 62(8): 937–943. <doi:10.1111/ped.14224>. <https://www.ncbi.nlm.nih.gov/pubmed/32153091>

Sakai T., Nomura Y., Sawai T., Hamada R., Gotoh Y., Yamamoto K., Ichioka S., Masuda T., Maruo Y., Honda M. Uptake of further investigations following universal urinary screening among elementary and junior high school students in Shiga Prefecture, Japan: A retrospective cohort study. *Nephrology (Carlton)* 2020; 25(8): 599–606. <doi:10.1111/nep.13710>. <https://www.ncbi.nlm.nih.gov/pubmed/32147900>

Yamada M., Uehara T., Suzuki H., Takenouchi T., Inui A., Ikemiyagi M., Kamimaki I., Kosaki K. Shortfall of exome analysis for diagnosis of Shwachman–Diamond syndrome: Mismapping due to the pseudogene SBDSP1. *Am J Med Genet A* 2020; 182(7): 1631–1636. <doi:10.1002/ajmg.a.61598>. <https://www.ncbi.nlm.nih.gov/pubmed/32412173>

<先天代謝異常、内分泌疾患>

Fujioka M., Akiyama T., Hosoya M., Kikuchi K., Fujiki Y., Saito Y., Yoshihama K., Ozawa H., Tsukada K., Nishio S. Y., Usami S. I., Matsunaga T., Hasegawa T., Sato Y., Ogawa K. A phase I/IIa double blind single institute trial of low dose sirolimus for Pendred

syndrome/DFNB4. *Medicine (Baltimore)* 2020; 99(19): e19763.

<doi:10.1097/MD.00000000000019763>.

<https://www.ncbi.nlm.nih.gov/pubmed/32384426>

Fukaishi T., Minami I., Masuda S., Miyachi Y., Tsujimoto K., Izumiyama H., Hashimoto K., Yoshida M., Takahashi S., Kashimada K., Morio T., Kosaki K., Maezawa Y., Yokote K., Yoshimoto T., Yamada T. A case of generalized lipodystrophy-associated progeroid syndrome treated by leptin replacement with short and long-term monitoring of the metabolic and endocrine profiles. *Endocr J* 2020; 67(2): 211–218. <doi:10.1507/endocrj.EJ19-0226>.

<https://www.ncbi.nlm.nih.gov/pubmed/31708526>

Hamada J., Ochi F., Sei Y., Takemoto K., Hirai H., Honda M., Shibata H., Hasegawa T., Eguchi M. A novel SOX10 variant in a Japanese girl with Waardenburg syndrome type 4C and Kallmann syndrome. *Hum Genome Var* 2020; 730. <doi:10.1038/s41439-020-00118-6>.

<https://www.ncbi.nlm.nih.gov/pubmed/33082981>

Hashimoto N., Dateki S., Suzuki E., Tsuchihashi T., Isobe A., Banno S., Kageyama T., Maeda N., Hatabu N., Sato R., Miharuru M., Fujita H., Komiyama O., Shimizu H., Hasegawa T., Yamazawa K. Compound heterozygous variants in the ABCG8 gene in a Japanese girl with sitosterolemia. *Hum Genome Var* 2020; 725. <doi:10.1038/s41439-020-00112-y>.

<https://www.ncbi.nlm.nih.gov/pubmed/33014402>

Hwang I. T., Mizuno Y., Amano N., Lee H. J., Shim Y. S., Nam H. K., Rhie Y. J., Yang S., Lee K. H., Hasegawa T., Kang M. J. Role of NPR2 mutation in idiopathic short stature: Identification of two novel mutations. *Mol Genet Genomic Med* 2020; 8(3): e1146.

<doi:10.1002/mgg3.1146>.

<https://www.ncbi.nlm.nih.gov/pubmed/31960617>

Ishii T., Tajima T., Kashimada K., Mukai T., Tanahashi Y., Katsumata N., Kanno J., Hamajima T., Miyako K., Ida S., Hasegawa T. Clinical Features of 57 Patients with Lipoid Congenital Adrenal Hyperplasia: Criteria for Nonclassic Form Revisited. *J Clin Endocrinol Metab* 2020; 105(11). <doi:10.1210/clinem/dgaa557>.

<https://www.ncbi.nlm.nih.gov/pubmed/32835366>

Iwahashi-Odano M., Nagasaki K., Fukami M., Nishioka J., Yatsuga S., Asakura Y., Adachi M., Muroya K., Hasegawa T., Narumi S. Congenital Hypothyroidism Due to Truncating

PAX8 Mutations: A Case Series and Molecular Function Studies. *J Clin Endocrinol Metab* 2020; 105(11). <doi:10.1210/clinem/dgaa584>.

<https://www.ncbi.nlm.nih.gov/pubmed/32841355>

Kagami R., Sato T., Ishii T., Araki E., Yamashita Y., Shibata H., Ishihara J., Hasegawa T. Central precocious puberty in a boy with pseudohypoparathyroidism type Ia due to a novel GNAS mutation. *Clin Pediatr Endocrinol* 2020; 29(2): 89–90. <doi:10.1297/cpe.29.89>.

<https://www.ncbi.nlm.nih.gov/pubmed/32313379>

Matsumoto R., Suga H., Aoi T., Bando H., Fukuoka H., Iguchi G., Narumi S., Hasegawa T., Muguruma K., Ogawa W., Takahashi Y. Congenital pituitary hypoplasia model demonstrates hypothalamic OTX2 regulation of pituitary progenitor cells. *J Clin Invest* 2020; 130(2): 641–654. <doi:10.1172/JCI127378>.

<https://www.ncbi.nlm.nih.gov/pubmed/31845906>

Matsuoka K., Sato Y., Hoshi S., Koguchi T., Ogawa S., Ishii T., Haga N., Hasegawa T., Kojima Y. Congenital lipoid adrenal hyperplasia: Immunohistochemical study of testosterone synthesis in Leydig cells. *IJU Case Rep* 2020; 3(2): 53–56. <doi:10.1002/iju5.12142>.

<https://www.ncbi.nlm.nih.gov/pubmed/32743469>

Nagasaki K., Takase K., Numakura C., Homma K., Hasegawa T., Fukami M. Foetal virilisation caused by overproduction of non-aromatisable 11-oxygenated C19 steroids in maternal adrenal tumour. *Hum Reprod* 2020; 35(11): 2609–2612.

<doi:10.1093/humrep/deaa221>.

<https://www.ncbi.nlm.nih.gov/pubmed/32862221>

Saito-Abe M., Yamamoto-Hanada K., Nakayama S. F., Hashimoto Y., Natsume O., Fukami M., Hasegawa T., Ohya Y. Reference values for salivary cortisol in healthy young infants by liquid chromatography–tandem mass spectrometry. *Pediatr Int* 2020; 62(7): 785–788.

<doi:10.1111/ped.14166>.

<https://www.ncbi.nlm.nih.gov/pubmed/31976606>

Sato T., Ishii T., Yamaguchi Y., Ichihashi Y., Ochiai D., Asanuma H., Kuroda T., Hasegawa T. Case Report: Prenatal Genetic Counseling to Parents of Fetuses Suspected of Having Ambiguous Genitalia. *Front Pediatr* 2020; 8569548. <doi:10.3389/fped.2020.569548>.

<https://www.ncbi.nlm.nih.gov/pubmed/33520882>

Sato T., Kusakawa M., Ichihashi Y., Ishii T., Hasegawa T. Testosterone priming increased growth hormone peak levels in the stimulation test and suppressed gonadotropin secretion in three Japanese adolescent boys. *Clin Pediatr Endocrinol* 2020; 29(3): 119–121.
<doi:10.1297/cpe.29.119>.

<https://www.ncbi.nlm.nih.gov/pubmed/32694889>

Uchida N., Ohnishi T., Kojima T., Takahashi T., Makita Y., Fukami M., Shibata H., Hasegawa T., Ishii T. Relapsing 6q24-related transient neonatal diabetes mellitus with insulin resistance: A case report. *Clin Pediatr Endocrinol* 2020; 29(4): 179–182.
<doi:10.1297/cpe.29.179>.

<https://www.ncbi.nlm.nih.gov/pubmed/33088017>

Uchida N., Shibata H., Nishimura G., Hasegawa T. A novel mutation in the ACAN gene in a family with autosomal dominant short stature and intervertebral disc disease. *Hum Genome Var* 2020; 7(1): 44. <doi:10.1038/s41439-020-00132-8>.

<https://www.ncbi.nlm.nih.gov/pubmed/33298914>

Yaginuma M., Sato T., Yuki K., Hasegawa T. Infant with trisomy 13 who developed acute elevation of intraocular pressure and glaucoma. *Congenit Anom (Kyoto)* 2020; 60(5): 151–152. <doi:10.1111/cga.12367>.

<https://www.ncbi.nlm.nih.gov/pubmed/31981267>

Yatsuga S., Amano N., Nakamura-Utsunomiya A., Kobayashi H., Takasawa K., Nagasaki K., Nakamura A., Nishigaki S., Numakura C., Fujiwara I., Minamitani K., Hasegawa T., Tajima T. Clinical characteristics of cytochrome P450 oxidoreductase deficiency: a nationwide survey in Japan. *Endocr J* 2020; 67(8): 853–857. <doi:10.1507/endocrj.EJ20-0011>.

<https://www.ncbi.nlm.nih.gov/pubmed/32321882>

<免疫異常、膠原病、リウマチ性疾患、感染症>

Arai S., Yamanaka T., Kasai M., Fukuyama T., Aizawa Y., Matsui K., Sato M., Matsui H., Saitoh A. Parechovirus-A3 encephalitis presenting with focal seizure mimicking herpes simplex virus infection. *J Infect Chemother* 2020; 26(7): 736–740.
<doi:10.1016/j.jiac.2020.02.003>.

<https://www.ncbi.nlm.nih.gov/pubmed/32201195>

Azekawa S., Namkoong H., Mitamura K., Kawaoka Y., Saito F. Co-infection with SARS-CoV-2 and influenza A virus. *IDCases* 2020; 20e00775. <doi:10.1016/j.idcr.2020.e00775>. <https://www.ncbi.nlm.nih.gov/pubmed/32368495>

Fujimori K., Yamada M., Maekawa T., Yotani N., Tamura E. I., Imadome K. I., Kubota M., Ishiguro A. A case of neonatal cytomegalovirus infection with severe thrombocytopenia that was successfully managed with empiric antiviral therapy. *IDCases* 2020; 19e00675. <doi:10.1016/j.idcr.2019.e00675>. <https://www.ncbi.nlm.nih.gov/pubmed/32226764>

Funaki T., Fukuda A., Sakamoto S., Kasahara M., Saitoh A., Miyairi I. Serostatus following polio-containing vaccination before and after liver transplantation. *Pediatr Transplant* 2020; 24(6): e13766. <doi:10.1111/petr.13766>. <https://www.ncbi.nlm.nih.gov/pubmed/32558028>

Funaki T., Shoji K., Fukuda A., Sakamoto S., Kasahara M., Miyairi I. Safety of LAVs administered after pediatric LT. *Pediatr Transplant* 2020e13937. <doi:10.1111/petr.13937>. <https://www.ncbi.nlm.nih.gov/pubmed/33314516>

Gu Y., Fujitomo Y., Soeda H., Nakahama C., Hasegawa N., Maesaki S., Maeda M., Matsumoto T., Miyairi I., Ohmagari N. A nationwide questionnaire survey of clinic doctors on antimicrobial stewardship in Japan. *J Infect Chemother* 2020; 26(2): 149–156. <doi:10.1016/j.jiac.2019.12.005>. <https://www.ncbi.nlm.nih.gov/pubmed/31879188>

Hosoba R., Makita S., Shiotsuka M., Kobayashi O., Nakano K., Muroya M., Okada N., Suzuki M., Ida H., Fukuhara S., Munakata W., Suzuki T., Maruyama D., Maeshima A. M., Matsushita H., Yamamoto N., Ohe Y., Iwata S., Izutsu K. COVID-19 pneumonia in a patient with adult T-cell leukemia-lymphoma. *J Clin Exp Hematop* 2020; 60(4): 174–178. <doi:10.3960/jslrt.20030>. <https://www.ncbi.nlm.nih.gov/pubmed/32879154>

Imamura T., Shoji K., Kono N., Kubota M., Nishimura N., Ishiguro A., Miyairi I. Allele frequencies of Bordetella pertussis virulence-associated genes identified from pediatric patients with severe respiratory infections. *J Infect Chemother* 2020; 26(7): 765–768. <doi:10.1016/j.jiac.2020.02.016>.

<https://www.ncbi.nlm.nih.gov/pubmed/32334951>

Kamei K., Miyairi I., Ishikura K., Ogura M., Shoji K., Arai K., Ito R., Kawai T., Ito S. Prospective study of live attenuated vaccines for patients receiving immunosuppressive agents. *PLoS One* 2020; 15(10): e0240217. <doi:10.1371/journal.pone.0240217>.

<https://www.ncbi.nlm.nih.gov/pubmed/33002085>

Kimura N., Ohnishi T., Sato S., Uejima Y., Suganuma E. Immunoglobulin A vasculitis with intramuscular hemorrhage: a case report. *Pediatr Int* 2020; 62(11): 1292–1294. <doi:10.1111/ped.14329>.

<https://www.ncbi.nlm.nih.gov/pubmed/33063916>

Kinoshita N., Komura M., Tsuzuki S., Shoji K., Miyairi I. The effect of preauthorization and prospective audit and feedback system on oral antimicrobial prescription for outpatients at a children's hospital in Japan. *J Infect Chemother* 2020; 26(6): 582–587. <doi:10.1016/j.jiac.2020.01.013>.

<https://www.ncbi.nlm.nih.gov/pubmed/32088130>

Kobari S., Kusakabe T., Momota M., Shibahara T., Hayashi T., Ozasa K., Morita H., Matsumoto K., Saito H., Ito S., Kuroda E., Ishii K. J. IL-33 Is Essential for Adjuvant Effect of Hydroxypropyl-beta-Cyclodextrin on the Protective Intranasal Influenza Vaccination. *Front Immunol* 2020; 11360. <doi:10.3389/fimmu.2020.00360>.

<https://www.ncbi.nlm.nih.gov/pubmed/32210964>

Kondo T., Okabayashi K., Sugiura K., Obara H., Takeuchi H., Wada N., Takano Y., Iwata S., Hasegawa N., Kitagawa Y. Effectiveness of active nasal surveillance culture for Methicillin-resistant *Staphylococcus aureus* in patients undergoing colorectal surgery. *J Infect Chemother* 2020; 26(12): 1244–1248. <doi:10.1016/j.jiac.2020.06.013>.

<https://www.ncbi.nlm.nih.gov/pubmed/32861579>

Louis K., Macedo C., Bailly E., Lau L., Ramaswami B., Marrari M., Landsittel D., Chang A., Chandran U., Fadakar P., Yamada M., Chalasani G., Randhawa P., Zeevi A., Singh H., Lefaucheur C., Metes D. Coordinated Circulating T Follicular Helper and Activated B Cell Responses Underlie the Onset of Antibody-Mediated Rejection in Kidney Transplantation. *J Am Soc Nephrol* 2020; 31(10): 2457–2474. <doi:10.1681/ASN.2020030320>.

<https://www.ncbi.nlm.nih.gov/pubmed/32723838>

Mitake H., Yasuhara A., Lopes T. J. S., Tagawa-Sakai Y., Shimizu K., Ozawa H., Kawakami C., Morikawa S., Sugaya N., Watanabe T., Kawaoka Y. Comparison of the Pathogenicity in Mice of A(H1N1)pdm09 Viruses Isolated between 2009 and 2015 in Japan. *Viruses* 2020; 12(2). <doi:10.3390/v12020155>.

<https://www.ncbi.nlm.nih.gov/pubmed/32013144>

Mitamura K., Shimizu H., Yamazaki M., Ichikawa M., Abe T., Yasumi Y., Ichikawa Y., Shibata T., Yoshihara M., Shiozaki K., Baba S., Kudo Y., Tokushima M., Konomi Y., Kawakami C. Clinical evaluation of ID NOW influenza A & B 2, a rapid influenza virus detection kit using isothermal nucleic acid amplification technology – A comparison with currently available tests. *J Infect Chemother* 2020; 26(2): 216–221.

<doi:10.1016/j.jiac.2019.08.015>.

<https://www.ncbi.nlm.nih.gov/pubmed/31558351>

Miyairi I., Shoji K., Kinoshita N., Saitoh J., Sugahara Y., Watanabe Y., Komura M., Kasai M., Horikoshi Y., Shinjoh M., Igarashi T., Pediatric Infection Control Network for the Japanese Association of Children's Hospitals Related Institutions. Prospective monitoring of carbapenem use and pseudomonal resistance across pediatric institutions. *Infect Control Hosp Epidemiol* 2020; 41(9): 1042–1047. <doi:10.1017/ice.2020.234>.

<https://www.ncbi.nlm.nih.gov/pubmed/32484118>

Morozumi M., Tajima T., Sakuma M., Shouji M., Meguro H., Saito K., Iwata S., Ubukata K. Sequence Type Changes Associated with Decreasing Macrolide-Resistant Mycoplasma pneumoniae, Japan. *Emerg Infect Dis* 2020; 26(9): 2210–2213.

<doi:10.3201/eid2609.191575>.

<https://www.ncbi.nlm.nih.gov/pubmed/32818419>

Noel K. C., Papenburg J., Lacroix J., Quach C., O'Donnell S., Gonzales M., Willson D. F., Gilfoyle E., McNally J. D., Reynolds S., Kazzaz Y., Kawaguchi A., Sato M., Kongkiattikul L., Leteurtre S., Dubos F., Karaca Y., Chiusolo F., Piva J., Dendukuri N., Fontela P. S., Canadian Critical Care Trials Group, the Pediatric Lung Injury, Sepsis Investigators Network. International Survey on Determinants of Antibiotic Duration and Discontinuation in Pediatric Critically Ill Patients. *Pediatr Crit Care Med* 2020; 21(9): e696–e706.

<doi:10.1097/PCC.0000000000002397>.

<https://www.ncbi.nlm.nih.gov/pubmed/32639469>

Ohnishi T., Asato S., Maeda A., Nagata M., Uwamino Y., Kamimaki I. Does the patient truly not have congenital anomalies of the kidney and urinary tract? Urinary tract infection caused by *Gardnerella vaginalis*. *Pediatr Int* 2020; 62(8): 1009–1010. <doi:10.1111/ped.14328>. <https://www.ncbi.nlm.nih.gov/pubmed/32851758>

Ohnishi T., Kamimaki I., Kobayashi R., Nakatogawa K., Amemiya A., Mishima Y., Asato S., Shikoro N., Nakazawa M. Verification of blood volume for blood culture and detection rate in pediatrics. *J Infect Chemother* 2020; 26(5): 471–474. <doi:10.1016/j.jiac.2019.12.008>. <https://www.ncbi.nlm.nih.gov/pubmed/31899078>

Ohnishi T., Mishima Y., Takizawa S., Tsutsumi K., Amemiya A., Akiyama N., Kanna Y., Asato S., Tomita M., Ikemiyagi M., Shikoro N., Nakazawa M., Kurihara N., Kamimaki I. Clinical Features of Febrile Urinary Tract Infection Caused by Extended-spectrum Beta-lactamase-producing *Escherichia Coli* in Children. *Keio J Med* 2020; 69(2): 43–47. <doi:10.2302/kjm.2019-0005-OA>. <https://www.ncbi.nlm.nih.gov/pubmed/31474677>

Okubo Y., Michihata N., Uda K., Kinoshita N., Horikoshi Y., Miyairi I. Impacts of Primary Care Physician System on Healthcare Utilization and Antibiotic Prescription: Difference-in-Differences and Causal Mediation Analyses. *Pediatr Infect Dis J* 2020; 39(10): 937–942. <doi:10.1097/INF.0000000000002762>. <https://www.ncbi.nlm.nih.gov/pubmed/32502123>

Okubo Y., Uda K., Kinoshita N., Horikoshi Y., Miyairi I., Michihata N., Matsui H., Fushimi K., Yasunaga H. National trends in appropriate antibiotics use among pediatric inpatients with uncomplicated lower respiratory tract infections in Japan. *J Infect Chemother* 2020; 26(11): 1122–1128. <doi:10.1016/j.jiac.2020.04.025>. <https://www.ncbi.nlm.nih.gov/pubmed/32792248>

Orimo K., Saito H., Matsumoto K., Morita H. Innate Lymphoid Cells in the Airways: Their Functions and Regulators. *Allergy Asthma Immunol Res* 2020; 12(3): 381–398. <doi:10.4168/aair.2020.12.3.381>. <https://www.ncbi.nlm.nih.gov/pubmed/32141254>

Osumi T., Yoshimura S., Sako M., Uchiyama T., Ishikawa T., Kawai T., Inoue E., Takimoto

T., Takeuchi I., Yamada M., Sakamoto K., Yoshida K., Kimura Y., Matsukawa Y., Matsumoto K., Imadome K. I., Arai K., Deguchi T., Imai K., Yuza Y., Matsumoto K., Onodera M., Kanegane H., Tomizawa D., Kato M. Prospective Study of Allogeneic Hematopoietic Stem Cell Transplantation with Post-Transplantation Cyclophosphamide and Antithymocyte Globulin from HLA-Mismatched Related Donors for Nonmalignant Diseases. *Biol Blood Marrow Transplant* 2020; 26(11): e286–e291. <doi:10.1016/j.bbmt.2020.08.008>. <https://www.ncbi.nlm.nih.gov/pubmed/32798657>

Radzikowska U., Ding M., Tan G., Zhakparov D., Peng Y., Wawrzyniak P., Wang M., Li S., Morita H., Altunbulakli C., Reiger M., Neumann A. U., Lunjani N., Traidl-Hoffmann C., Nadeau K. C., O'Mahony L., Akdis C., Sokolowska M. Distribution of ACE2, CD147, CD26, and other SARS-CoV-2 associated molecules in tissues and immune cells in health and in asthma, COPD, obesity, hypertension, and COVID-19 risk factors. *Allergy* 2020; 75(11): 2829–2845. <doi:10.1111/all.14429>. <https://www.ncbi.nlm.nih.gov/pubmed/32496587>

Saito J., Shoji K., Oho Y., Aoki S., Matsumoto S., Yoshida M., Nakamura H., Kaneko Y., Hayashi T., Yamatani A., Capparelli E., Miyairi I. Meropenem pharmacokinetics during extracorporeal membrane oxygenation and continuous haemodialysis: a case report. *J Glob Antimicrob Resist* 2020; 22651–655. <doi:10.1016/j.jgar.2020.04.029>. <https://www.ncbi.nlm.nih.gov/pubmed/32417590>

Saitoh A., Saitoh A., Katsuta T., Mine M., Kamiya H., Miyairi I., Ishiwada N., Oshiro M., Kira R., Shimizu N., Suga S., Tsugawa T., Fujioka M., Miyazaki C., Morioka I., Korematsu S., Nakano T., Tanaka-Taya K., Yoshikawa T., Iwata S., Kusuhara K., Azuma H., Moriuchi H., Okabe N., Hosoya M., Tsutsumi H., Okada K. Effect of a vaccine information statement (VIS) on immunization status and parental knowledge, attitudes, and beliefs regarding infant immunization in Japan. *Vaccine* 2020; 38(50): 8049–8054. <doi:10.1016/j.vaccine.2020.10.049>. <https://www.ncbi.nlm.nih.gov/pubmed/33139133>

Sakamoto A., Yamada M., Tsujimoto S. I., Osumi T., Arai K., Tomizawa D., Ishiguro A., Matsumoto K., Imadome K. I., Kato M. A case of human herpesvirus 6 encephalitis following pediatric hematopoietic stem cell transplantation: early diagnosis and treatment matters. *Int J Hematol* 2020; 112(5): 751–754. <doi:10.1007/s12185-020-02905-x>. <https://www.ncbi.nlm.nih.gov/pubmed/32529583>

Sasaki J., Shiino Y., Kato Y., Kudo D., Fujita M., Miyairi I., Mochizuki T., Okuda H., Nagato T., Nabetani Y., Takahashi T., Committee for Infection Control in the Emergency Department Joint Working Group. Checklist for infection control in the emergency department. *Acute Med Surg* 2020; 7(1): e540. <doi:10.1002/ams2.540>.

<https://www.ncbi.nlm.nih.gov/pubmed/33364033>

Sato S., Ohnishi T., Uejima Y., Furuichi M., Fujinaga S., Imai K., Nakamura K., Kawano Y., Suganuma E. Induction therapy with rituximab for lupus nephritis due to prolidase deficiency. *Rheumatology (Oxford)* 2020; 59(10): e57–e59. <doi:10.1093/rheumatology/keaa051>.

<https://www.ncbi.nlm.nih.gov/pubmed/32107546>

Seki Y., Oda Y., Sugaya N. Very high sensitivity of a rapid influenza diagnostic test in adults and elderly individuals within 48 hours of the onset of illness. *PLoS One* 2020; 15(5): e0231217. <doi:10.1371/journal.pone.0231217>.

<https://www.ncbi.nlm.nih.gov/pubmed/32374728>

Shinjoh M., Yamaguchi Y., Furuichi M., Yaginuma M., Takahashi T., Iwata S. Recent trends in pediatric bacterial meningitis in Japan, 2016–2018 – *S. agalactiae* has been the most common pathogen. *J Infect Chemother* 2020; 26(10): 1033–1041.

<doi:10.1016/j.jiac.2020.05.018>.

<https://www.ncbi.nlm.nih.gov/pubmed/32546331>

Shoji K., Michihata N., Miyairi I., Matsui H., Fushimi K., Yasunaga H. Recent epidemiology of *Pneumocystis pneumonia* in Japan. *J Infect Chemother* 2020; 26(12): 1260–1264.

<doi:10.1016/j.jiac.2020.07.006>.

<https://www.ncbi.nlm.nih.gov/pubmed/32753118>

Sokolowska M., Lukasik Z. M., Agache I., Akdis C. A., Akdis D., Akdis M., Barcik W., Brough H. A., Eiwegger T., Eljaszewicz A., Eyerich S., Feleszko W., Gomez-Casado C., Hoffmann-Sommergruber K., Janda J., Jimenez-Saiz R., Jutel M., Knol E. F., Kortekaas Krohn I., Kothari A., Makowska J., Moniuszko M., Morita H., O'Mahony L., Nadeau K., Ozdemir C., Pali-Scholl I., Palomares O., Papaleo F., Prunicki M., Schmidt-Weber C. B., Sediva A., Schwarze J., Shamji M. H., Tramper-Stranders G. A., van de Veen W., Untersmayr E. Immunology of COVID-19: Mechanisms, clinical outcome, diagnostics, and perspectives—A report of the European Academy of Allergy and Clinical Immunology

(EAACI). *Allergy* 2020; 75(10): 2445–2476. <doi:10.1111/all.14462>.

<https://www.ncbi.nlm.nih.gov/pubmed/32584441>

Sugihara E., Hashimoto N., Osuka S., Shimizu T., Ueno S., Okazaki S., Yaguchi T., Kawakami Y., Kosaki K., Sato T. A., Okamoto S., Saya H. The Inhibitor of Apoptosis Protein Livin Confers Resistance to Fas-Mediated Immune Cytotoxicity in Refractory Lymphoma. *Cancer Res* 2020; 80(20): 4439–4450. <doi:10.1158/0008-5472.CAN-19-3993>.

<https://www.ncbi.nlm.nih.gov/pubmed/32928920>

Suzuki J., Kobayashi S., Yoshida N., Azuma Y., Kobayashi-Ogata N., Kartikasari D. P., Yanagawa Y., Iwata S. Phylogenetic position of *Nyctotherus teleacus* isolated from a tortoise (*Astrochelys radiata*) and its electron microscopic features. *J Vet Med Sci* 2020; 82(6): 699–703. <doi:10.1292/jvms.20-0004>.

<https://www.ncbi.nlm.nih.gov/pubmed/32336700>

Takashita E., Abe T., Morita H., Nagata S., Fujisaki S., Miura H., Shirakura M., Kishida N., Nakamura K., Kuwahara T., Mitamura K., Ichikawa M., Yamazaki M., Watanabe S., Hasegawa H., Influenza Virus Surveillance Group of Japan. Influenza A(H1N1)pdm09 virus exhibiting reduced susceptibility to baloxavir due to a PA E23K substitution detected from a child without baloxavir treatment. *Antiviral Res* 2020; 180104828. <doi:10.1016/j.antiviral.2020.104828>.

<https://www.ncbi.nlm.nih.gov/pubmed/32574689>

Tamaoka S., Shindo J., Tsuchihashi T., Bamba M., Iwata S. A 9-year-old boy with a sinus-related epidural abscess caused by *Listeria monocytogenes*. *Pediatr Int* 2020; 62(4): 502–503. <doi:10.1111/ped.14111>.

<https://www.ncbi.nlm.nih.gov/pubmed/32307827>

Taniyama Y., Shoji K., Hashimoto M., Ishiguro A., Miyairi I. Impact of the 3-day rule for stool culture in a children's hospital. *Pediatr Int* 2020; 62(2): 246–247. <doi:10.1111/ped.14100>.

<https://www.ncbi.nlm.nih.gov/pubmed/32104991>

Terada A., Ohnishi T., Mishima Y., Akiyama N., Kanna Y., Asato S., Tomita M., Ikemiyagi M., Shikoro N., Nakazawa M., Kurihara N., Tado M., Yachie A., Kamimaki I. One-month-old boy with group B streptococcal meningitis, subdural effusion, and high levels of interleukin-6.

J Infect Chemother 2020; 26(10): 1090–1094. <doi:10.1016/j.jiac.2020.06.017>.

<https://www.ncbi.nlm.nih.gov/pubmed/32646818>

Ubukata K., Wajima T., Morozumi M., Sakuma M., Tajima T., Matsubara K., Itahashi K., Iwata S. Changes in epidemiologic characteristics and antimicrobial resistance of *Streptococcus pyogenes* isolated over 10 years from Japanese children with pharyngotonsillitis. *J Med Microbiol* 2020; 69(3): 443–450. <doi:10.1099/jmm.0.001158>.

<https://www.ncbi.nlm.nih.gov/pubmed/32011228>

Uda K., Funaki T., Shoji K., Kato A., Miyairi I. High proportion of multidrug-resistant organisms in children hospitalized abroad. *Am J Infect Control* 2020; 48(5): 578–580. <doi:10.1016/j.ajic.2019.08.013>.

<https://www.ncbi.nlm.nih.gov/pubmed/31519478>

Uda K., Uehara Y., Morimoto Y., Hiramatsu K., Miyairi I. A Pediatric Case of Septic Arthritis Caused by Methicillin-Resistant *Staphylococcus aureus* with Pantone-Valentine Leukocidin and Toxic Shock Syndrome Toxin-1. *Jpn J Infect Dis* 2020; 73(3): 259–262. <doi:10.7883/yoken.JJID.2019.436>.

<https://www.ncbi.nlm.nih.gov/pubmed/32350217>

Uehara T., Hayden F. G., Kawaguchi K., Omoto S., Hurt A. C., De Jong M. D., Hirotsu N., Sugaya N., Lee N., Baba K., Shishido T., Tsuchiya K., Portsmouth S., Kida H. Treatment-Emergent Influenza Variant Viruses With Reduced Baloxavir Susceptibility: Impact on Clinical and Virologic Outcomes in Uncomplicated Influenza. *J Infect Dis* 2020; 221(3): 346–355. <doi:10.1093/infdis/jiz244>.

<https://www.ncbi.nlm.nih.gov/pubmed/31309975>

Yaginuma M., Sato T., Yuki K., Hasegawa T. Infant with trisomy 13 who developed acute elevation of intraocular pressure and glaucoma. *Congenit Anom (Kyoto)* 2020; 60(5): 151–152. <doi:10.1111/cga.12367>.

<https://www.ncbi.nlm.nih.gov/pubmed/31981267>

Yamada Masaki, Nowalk Andrew, Green Michael. in *Emerging Transplant Infections: Clinical Challenges and Implications* (eds Michele I. Morris, Camille Nelson Kotton, & Camille Wolfe) 1–29 (Springer International Publishing, 2020).

Yamada M., Pellett Madan R. CMV as culprit or bystander: The debate continues. *Pediatr Transplant* 2020; 24(8): e13865. <doi:10.1111/ptr.13865>.

<https://www.ncbi.nlm.nih.gov/pubmed/33044777>

Yamazawa E., Ohno M., Satomi K., Yoshida A., Miyakita Y., Takahashi M., Satomi N., Asanome T., Maeshima A., Shiotsuka M., Iwata S., Yamasaki H., Morishima Y., Sugiyama H., Narita Y. First case of human neurocoenurosis caused by *Taenia serialis*: A case report. *Int J Infect Dis* 2020; 92:171–174. <doi:10.1016/j.ijid.2020.01.004>.

<https://www.ncbi.nlm.nih.gov/pubmed/31927059>

Yoshida M., Takeuchi I., Shoji K., Miyairi I., Arai K. Bacillus Calmette–Guerin Cervical Lymphadenitis in a 6-Year-Old Boy on Infliximab for Inflammatory Bowel Disease. *Pediatr Infect Dis J* 2020; 39(9): e242–e244. <doi:10.1097/INF.0000000000002712>.

<https://www.ncbi.nlm.nih.gov/pubmed/32345827>